

Egbert Bakker

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

134
papers

9,051
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93
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139
ext. papers

9,746
ext. citations

8.7
avg, IF

5
L-index

#	Paper	IF	Citations
134	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. <i>Nature Genetics</i> , 2012 , 44, 1370-4	36.3	405
133	BRCA1 genomic deletions are major founder mutations in Dutch breast cancer patients. <i>Nature Genetics</i> , 1997 , 17, 341-5	36.3	379
132	Genome-wide association study identifies three loci associated with melanoma risk. <i>Nature Genetics</i> , 2009 , 41, 920-5	36.3	360
131	Clinical findings with implications for genetic testing in families with clustering of colorectal cancer. <i>New England Journal of Medicine</i> , 1998 , 339, 511-8	59.2	346
130	Rapid detection of BRCA1 mutations by the protein truncation test. <i>Nature Genetics</i> , 1995 , 10, 208-12	36.3	283
129	Guidelines for diagnostic next-generation sequencing. <i>European Journal of Human Genetics</i> , 2016 , 24, 2-5	5.3	268
128	Estimating Y chromosome specific microsatellite mutation frequencies using deep rooting pedigrees. <i>Human Molecular Genetics</i> , 1997 , 6, 799-803	5.6	218
127	Isolation of probes detecting restriction fragment length polymorphisms from X chromosome-specific libraries: potential use for diagnosis of Duchenne muscular dystrophy. <i>Human Genetics</i> , 1985 , 70, 148-56	6.3	215
126	Hereditary nonpolyposis colorectal cancer families not complying with the Amsterdam criteria show extremely low frequency of mismatch-repair-gene mutations. <i>American Journal of Human Genetics</i> , 1997 , 61, 329-35	11	204
125	Population-based incidence and prevalence of facioscapulohumeral dystrophy. <i>Neurology</i> , 2014 , 83, 1056-9	20.3	203
124	Genome-wide association study identifies three new melanoma susceptibility loci. <i>Nature Genetics</i> , 2011 , 43, 1108-13	36.3	203
123	Direct detection of more than 50% of the Duchenne muscular dystrophy mutations by field inversion gels. <i>Nature</i> , 1987 , 329, 640-2	50.4	191
122	Germline mosaicism and Duchenne muscular dystrophy mutations. <i>Nature</i> , 1987 , 329, 554-6	50.4	187
121	An extensive analysis of Y-chromosomal microsatellite haplotypes in globally dispersed human populations. <i>American Journal of Human Genetics</i> , 2001 , 68, 990-1018	11	167
120	Genome-wide association study identifies novel loci predisposing to cutaneous melanoma. <i>Human Molecular Genetics</i> , 2011 , 20, 5012-23	5.6	164
119	Germinal mosaicism increases the recurrence risk for new Duchenne muscular dystrophy mutations. <i>Journal of Medical Genetics</i> , 1989 , 26, 553-9	5.8	163
118	Array-CGH detection of micro rearrangements in mentally retarded individuals: clinical significance of imbalances present both in affected children and normal parents. <i>Journal of Medical Genetics</i> , 2006 , 43, 180-6	5.8	161

117	Comprehensive detection of genomic duplications and deletions in the DMD gene, by use of multiplex amplifiable probe hybridization. <i>American Journal of Human Genetics</i> , 2002 , 71, 365-74	11	155
116	Multiple osteochondromas: mutation update and description of the multiple osteochondromas mutation database (MOdb). <i>Human Mutation</i> , 2009 , 30, 1620-7	4.7	154
115	EXT-mutation analysis and loss of heterozygosity in sporadic and hereditary osteochondromas and secondary chondrosarcomas. <i>American Journal of Human Genetics</i> , 1999 , 65, 689-98	11	151
114	Mutations in the EXT1 and EXT2 genes in hereditary multiple exostoses. <i>American Journal of Human Genetics</i> , 1998 , 62, 346-54	11	149
113	A physical map of 4 million bp around the Duchenne muscular dystrophy gene on the human X-chromosome. <i>Cell</i> , 1986 , 47, 499-504	56.2	142
112	Two subsets of human alphoid repetitive DNA show distinct preferential localization in the pericentric regions of chromosomes 13, 18, and 21. <i>Cytogenetic and Genome Research</i> , 1986 , 41, 193-201 ^{1.9}	1.9	142
111	Two-color multiplex ligation-dependent probe amplification: detecting genomic rearrangements in hereditary multiple exostoses. <i>Human Mutation</i> , 2004 , 24, 86-92	4.7	136
110	The FSHD2 gene SMCHD1 is a modifier of disease severity in families affected by FSHD1. <i>American Journal of Human Genetics</i> , 2013 , 93, 744-51	11	126
109	Analysis of molecular variance (AMOVA) of Y-chromosome-specific microsatellites in two closely related human populations. <i>Human Molecular Genetics</i> , 1996 , 5, 1029-33	5.6	126
108	De novo facioscapulohumeral muscular dystrophy: frequent somatic mosaicism, sex-dependent phenotype, and the role of mitotic transchromosomal repeat interaction between chromosomes 4 and 10. <i>American Journal of Human Genetics</i> , 2000 , 66, 26-35	11	118
107	Diagnostic guidelines for high-resolution melting curve (HRM) analysis: an interlaboratory validation of BRCA1 mutation scanning using the 96-well LightScanner. <i>Human Mutation</i> , 2009 , 30, 899-909	4.7	116
106	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. <i>Human Molecular Genetics</i> , 2015 , 24, 659-69	5.6	106
105	A deletion hot spot in the Duchenne muscular dystrophy gene. <i>Genomics</i> , 1988 , 2, 101-8	4.3	106
104	A variant in FTO shows association with melanoma risk not due to BMI. <i>Nature Genetics</i> , 2013 , 45, 428-32, 432e1	36.3	95
103	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. <i>Human Genetics</i> , 1992 , 88, 249-57	6.3	92
102	Van Buchem disease: clinical, biochemical, and densitometric features of patients and disease carriers. <i>Journal of Bone and Mineral Research</i> , 2013 , 28, 848-54	6.3	89
101	Intronic variants in BRCA1 and BRCA2 that affect RNA splicing can be reliably selected by splice-site prediction programs. <i>Human Mutation</i> , 2009 , 30, 107-14	4.7	88
100	Melanocytic nevi, nevus genes, and melanoma risk in a large case-control study in the United Kingdom. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2010 , 19, 2043-54	4	83

99	Complete allele information in the diagnosis of facioscapulohumeral muscular dystrophy by triple DNA analysis. <i>Annals of Neurology</i> , 2001 , 50, 816-9	9.4	80
98	The FSHD-linked locus D4F104S1 (p13E-11) ON 4q35 has a homologue on 10qter. <i>Muscle and Nerve</i> , 1995 , 18, S39-S44	3.4	79
97	Localization of the polymorphic human calcitonin gene on chromosome 11. <i>Human Genetics</i> , 1984 , 66, 309-12	6.3	76
96	Dystrophin nonsense mutation induces different levels of exon 29 skipping and leads to variable phenotypes within one BMD family. <i>European Journal of Human Genetics</i> , 2000 , 8, 793-6	5.3	75
95	Long-range genomic map of the Duchenne muscular dystrophy (DMD) gene: isolation and use of J66 (DXS268), a distal intragenic marker. <i>Genomics</i> , 1987 , 1, 329-36	4.3	69
94	A new diagnostic workflow for patients with mental retardation and/or multiple congenital abnormalities: test arrays first. <i>European Journal of Human Genetics</i> , 2009 , 17, 1394-402	5.3	63
93	Ring chromosome formation as a novel escape mechanism in patients with inverted duplication and terminal deletion. <i>European Journal of Human Genetics</i> , 2007 , 15, 548-55	5.3	63
92	Zygoty determination in newborn twins using DNA variants. <i>Journal of Medical Genetics</i> , 1985 , 22, 279-82	5.3	63
91	Molecular diagnostics of Meckel-Gruber syndrome highlights phenotypic differences between MKS1 and MKS3. <i>Human Genetics</i> , 2007 , 121, 591-9	6.3	62
90	No haploinsufficiency but loss of heterozygosity for EXT in multiple osteochondromas. <i>American Journal of Pathology</i> , 2010 , 177, 1946-57	5.8	59
89	Fine mapping of the Huntington disease linked D4S10 locus by non-radioactive in situ hybridization. <i>Human Genetics</i> , 1986 , 73, 354-7	6.3	57
88	High resolution deletion breakpoint mapping in the DMD gene by whole cosmid hybridization. <i>Nucleic Acids Research</i> , 1989 , 17, 5611-21	20.1	56
87	Evolution of cardiac abnormalities in Becker muscular dystrophy over a 13-year period. <i>Journal of Neurology</i> , 1997 , 244, 657-63	5.5	55
86	Somatic mosaicism in FSHD often goes undetected. <i>Annals of Neurology</i> , 2004 , 55, 845-50	9.4	53
85	DNA probe analysis for carrier detection and prenatal diagnosis of Duchenne muscular dystrophy: a standard diagnostic procedure. <i>Journal of Medical Genetics</i> , 1986 , 23, 573-80	5.8	53
84	Keratosis Follicularis Spinulosa Decalvans is caused by mutations in MBTPS2. <i>Human Mutation</i> , 2010 , 31, 1125-33	4.7	52
83	The apolipoprotein E epsilon 4 allele does not influence the clinical expression of the amyloid precursor protein gene codon 693 or 692 mutations. <i>Annals of Neurology</i> , 1994 , 36, 434-7	9.4	52
82	Somatic origin of inherited haemophilia A. <i>Human Genetics</i> , 1990 , 85, 288-92	6.3	49

81	Pre- and postsynaptic neuromuscular junction abnormalities in musk myasthenia. <i>Muscle and Nerve</i> , 2010 , 42, 283-8	3.4	45
80	DGGE-based whole-gene mutation scanning of the dystrophin gene in Duchenne and Becker muscular dystrophy patients. <i>Human Mutation</i> , 2004 , 23, 57-66	4.7	45
79	Mutation screening of EXT1 and EXT2 by direct sequence analysis and MLPA in patients with multiple osteochondromas: splice site mutations and exonic deletions account for more than half of the mutations. <i>European Journal of Human Genetics</i> , 2005 , 13, 470-4	5.3	44
78	Genetic heterogeneity for Duchenne-like muscular dystrophy (DLMD) based on linkage and 50 DAG analysis. <i>Human Molecular Genetics</i> , 1993 , 2, 1945-7	5.6	44
77	Early onset facioscapulohumeral muscular dystrophy. <i>Muscle and Nerve</i> , 1995 , 18, S67-S72	3.4	44
76	Confirmation of the 2p locus for the mild autosomal recessive limb-girdle muscular dystrophy gene (LGMD2B) in three families allows refinement of the candidate region. <i>Genomics</i> , 1995 , 27, 192-5	4.3	39
75	Is the DNA sequence the gold standard in genetic testing? Quality of molecular genetic tests assessed. <i>Clinical Chemistry</i> , 2006 , 52, 557-8	5.5	38
74	The fragile X syndrome in a large family. III. Investigations on linkage of flanking DNA markers with the fragile site Xq27. <i>Journal of Medical Genetics</i> , 1987 , 24, 413-21	5.8	38
73	Development of additional RFLP probes near the locus for Duchenne muscular dystrophy by cosmid cloning of the DXS84 (754) locus. <i>Human Genetics</i> , 1986 , 74, 270-4	6.3	36
72	Single molecule sequencing of free DNA from maternal plasma for noninvasive trisomy 21 detection. <i>Clinical Chemistry</i> , 2012 , 58, 699-706	5.5	33
71	Genotype-phenotype correlations in 19 Dutch cases with APC gene deletions and a literature review. <i>European Journal of Human Genetics</i> , 2007 , 15, 1034-42	5.3	32
70	Fine-tiling array CGH to improve diagnostics for β and δ thalassemia rearrangements. <i>Human Mutation</i> , 2012 , 33, 272-80	4.7	31
69	Dystrophin analysis in carriers of Duchenne and Becker muscular dystrophy. <i>Neurology</i> , 2005 , 65, 1984-66.5		31
68	Novel Leptin Receptor Mutations Identified in Two Girls with Severe Obesity Are Associated with Increased Bone Mineral Density. <i>Hormone Research in Paediatrics</i> , 2016 , 85, 412-20	3.3	31
67	An urgent need for a change in policy revealed by a study on prenatal testing for Duchenne muscular dystrophy. <i>European Journal of Human Genetics</i> , 2013 , 21, 21-6	5.3	30
66	Non-invasive prenatal diagnosis of beta-thalassemia and sickle-cell disease using pyrophosphorolysis-activated polymerization and melting curve analysis. <i>Prenatal Diagnosis</i> , 2012 , 32, 578-87	3.2	28
65	Rapid and cost effective detection of small mutations in the DMD gene by high resolution melting curve analysis. <i>Neuromuscular Disorders</i> , 2009 , 19, 383-90	2.9	28
64	Alzheimer β disease and hereditary cerebral hemorrhage with amyloidosis-Dutch type share a decrease in cerebrospinal fluid levels of amyloid beta-protein precursor. <i>Annals of Neurology</i> , 1992 , 32, 215-8	9.4	27

63	Asn540Thr substitution in the fibroblast growth factor receptor 3 tyrosine kinase domain causing hypochondroplasia. <i>Human Mutation</i> , 1998 , Suppl 1, S62-5	4.7	26
62	A European pilot quality assessment scheme for molecular diagnosis of HuntingtonQ disease. <i>European Journal of Human Genetics</i> , 1999 , 7, 217-22	5.3	26
61	Unclassified variants in disease-causing genes: nonuniformity of genetic testing and counselling, a proposal for guidelines. <i>European Journal of Human Genetics</i> , 2005 , 13, 525-7	5.3	25
60	Hemizyosity for SMCHD1 in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. <i>Human Mutation</i> , 2015 , 36, 679-83	4.7	24
59	Y chromosome detection by Real Time PCR and pyrophosphorolysis-activated polymerisation using free fetal DNA isolated from maternal plasma. <i>Prenatal Diagnosis</i> , 2007 , 27, 932-7	3.2	24
58	Point mutation screening for 16 exons of the dystrophin gene by multiplex single-strand conformation polymorphism analysis. <i>Human Mutation</i> , 1995 , 5, 235-42	4.7	24
57	Molecular genetic analysis of two families with keratosis follicularis spinulosa decalvans: refinement of gene localization and evidence for genetic heterogeneity. <i>Human Genetics</i> , 1997 , 100, 520-4	6.3	23
56	Severe nonspecific X-linked mental retardation caused by a proximally Xp located gene: intragenic heterogeneity or a new form of X-linked mental retardation?. <i>American Journal of Medical Genetics Part A</i> , 1993 , 46, 172-5		22
55	No evidence of genetic heterogeneity in Brazilian facioscapulohumeral muscular dystrophy families (FSHD) with 4q markers. <i>Human Molecular Genetics</i> , 1993 , 2, 557-62	5.6	20
54	Additional cryptic CNVs in mentally retarded patients with apparently balanced karyotypes. <i>European Journal of Medical Genetics</i> , 2010 , 53, 227-33	2.6	19
53	Presymptomatic, prenatal, and exclusion testing for Huntington disease using seven closely linked DNA markers. <i>American Journal of Medical Genetics Part A</i> , 1991 , 39, 217-22		19
52	An unusual variant of Becker muscular dystrophy. <i>Annals of Neurology</i> , 1990 , 27, 578-81	9.4	19
51	A t(4;6)(q12;p23) translocation disrupts a membrane-associated O-acetyl transferase gene (MBOAT1) in a patient with a novel brachydactyly-syndactyly syndrome. <i>European Journal of Human Genetics</i> , 2007 , 15, 743-51	5.3	18
50	Two step procedure for early diagnosis of polycystic kidney disease with polymorphic DNA markers on both sides of the gene. <i>Journal of Medical Genetics</i> , 1990 , 27, 614-7	5.8	18
49	Isolation of a random cosmid clone, cX5, which defines a new polymorphic locus DXS148 near the locus for Duchenne muscular dystrophy. <i>Human Genetics</i> , 1986 , 74, 275-9	6.3	18
48	Genetic epidemiology and preventive healthcare in multiethnic societies: the hemoglobinopathies. <i>International Journal of Environmental Research and Public Health</i> , 2014 , 11, 6136-46	4.6	17
47	An Xp22.1-p22.2 YAC contig encompassing the disease loci for RS, KFSD, CLS, HYP and RP15: refined localization of RS. <i>European Journal of Human Genetics</i> , 1996 , 4, 101-4	5.3	17
46	Noninvasive prenatal diagnosis of Huntington disease: detection of the paternally inherited expanded CAG repeat in maternal plasma. <i>Prenatal Diagnosis</i> , 2015 , 35, 945-9	3.2	16

45	Search for copy number alterations in the MEFV gene using multiplex ligation probe amplification, experience from three diagnostic centres. <i>European Journal of Human Genetics</i> , 2008 , 16, 1404-6	5.3	16
44	Three new cases with a mosaicism involving a normal cell line and a cryptic unbalanced autosomal reciprocal translocation. <i>European Journal of Medical Genetics</i> , 2011 , 54, e409-12	2.6	15
43	Haplotypes encoding the factor VIII 1241 Glu variation, factor VIII levels and the risk of venous thrombosis. <i>Thrombosis and Haemostasis</i> , 2006 , 95, 942-8	7	15
42	New possibilities for prenatal diagnosis of muscular dystrophies: forced myogenesis with an adenoviral MyoD-vector. <i>Lancet, The</i> , 1999 , 353, 727-8	4.0	15
41	Immunological study of dystrophin in Duchenne fetus. <i>Lancet, The</i> , 1989 , 2, 1212-3	4.0	15
40	Candidate gene-based association study of antipsychotic-induced movement disorders in long-stay psychiatric patients: a prospective study. <i>PLoS ONE</i> , 2012 , 7, e36561	3.7	15
39	The jumping SHOX gene--crossover in the pseudoautosomal region resulting in unusual inheritance of Leri-Weill dyschondrosteosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011 , 96, E356-9	5.6	14
38	Identification of copy number variants associated with BPES-like phenotypes. <i>Human Genetics</i> , 2008 , 124, 489-98	6.3	14
37	Genome-wide linkage scan in Dutch hereditary non-BRCA1/2 breast cancer families identifies 9q21-22 as a putative breast cancer susceptibility locus. <i>Genes Chromosomes and Cancer</i> , 2008 , 47, 947-58	5	14
36	Evidence for the absence of intron H of the histidine-rich glycoprotein (HRG) gene: genetic mapping and in situ localization of HRG to chromosome 3q28-q29. <i>Genomics</i> , 1994 , 19, 195-7	4.3	14
35	The DMD gene analysed by field inversion gel electrophoresis. <i>British Medical Bulletin</i> , 1989 , 45, 644-58	5.4	14
34	An anonymous single copy chromosome 22 clone, D22S10 (22c1-18) identifies an RFLP with PstI. <i>Nucleic Acids Research</i> , 1985 , 13, 7167	20.1	14
33	Location on the Human Genetic Linkage Map of 26 Genes Involved in Blood Coagulation. <i>Thrombosis and Haemostasis</i> , 1997 , 77, 0873-0878	7	13
32	Toward health technology assessment of whole-genome sequencing diagnostic tests: challenges and solutions. <i>Personalized Medicine</i> , 2017 , 14, 235-247	2.2	12
31	Experiences with array-based sequence capture; toward clinical applications. <i>European Journal of Human Genetics</i> , 2011 , 19, 50-5	5.3	12
30	Denaturing and non-denaturing gel electrophoresis as methods for the detection of junctional diversity in rearranged T cell receptor sequences. <i>Journal of Immunological Methods</i> , 1995 , 181, 101-14	2.5	12
29	Phenotypic characterization of patients with deletions in the 3' flanking SHOX region. <i>PeerJ</i> , 2013 , 1, e35	3.1	12
28	A 400kb duplication, 2.4Mb triplication and 130kb duplication of 9q34.3 in a patient with severe mental retardation. <i>European Journal of Medical Genetics</i> , 2008 , 51, 479-87	2.6	11

27	Two additional RFLPs at the D4S10 locus, useful for Huntington ^Q disease (HD)-family studies. <i>Nucleic Acids Research</i> , 1987 , 15, 9100	20.1	11
26	A novel (Leu183Pro-)mutation in the HFE-gene co-inherited with the Cys282Tyr mutation in two unrelated Dutch hemochromatosis patients. <i>Blood Cells, Molecules, and Diseases</i> , 2008 , 40, 334-8	2.1	10
25	Somatic Triple Mosaicism in a Carrier of X-Linked Chronic Granulomatous Disease. <i>Blood</i> , 1998 , 91, 252-257		10
24	European registration process for Clinical Laboratory Geneticists in genetic healthcare. <i>European Journal of Human Genetics</i> , 2017 , 25, 515-519	5.3	9
23	Successful noninvasive trisomy 18 detection using single molecule sequencing. <i>Clinical Chemistry</i> , 2013 , 59, 705-9	5.5	9
22	Mrsf1a-pap, a novel methylation-based assay for the detection of cell-free fetal DNA in maternal plasma. <i>PLoS ONE</i> , 2013 , 8, e84051	3.7	9
21	First trimester prenatal diagnosis of haemophilia A: two years ^Q experience. <i>Prenatal Diagnosis</i> , 1988 , 8, 411-21	3.2	9
20	Mapping around the Xq13.1 breakpoints of two X/A translocations in hypohidrotic ectodermal dysplasia (EDA) female patients. <i>Genomics</i> , 1992 , 14, 523-5	4.3	8
19	An infrequent DNA polymorphism associated with severe von Willebrand ^Q disease. <i>British Journal of Haematology</i> , 1990 , 75, 78-81	4.5	8
18	Development of a comprehensive noninvasive prenatal test. <i>Genetics and Molecular Biology</i> , 2018 , 41, 545-554	2	7
17	A novel keratin 13 variant in a four-generation family with white sponge nevus. <i>Clinical Case Reports (discontinued)</i> , 2017 , 5, 1503-1509	0.7	7
16	A Novel Targeted Approach for Noninvasive Detection of Paternally Inherited Mutations in Maternal Plasma. <i>Journal of Molecular Diagnostics</i> , 2015 , 17, 590-6	5.1	6
15	The Dutch Uniform Multicenter Registration system for genetic disorders and malformation syndromes. <i>American Journal of Medical Genetics Part A</i> , 1997 , 70, 444-7		6
14	Critical points for an accurate human genome analysis. <i>Human Mutation</i> , 2017 , 38, 912-921	4.7	5
13	Molecular spectrum of β globin gene defects in the Omani population. <i>Hemoglobin</i> , 2014 , 38, 422-6	0.6	5
12	Known and new β globin gene mutations and other factors influencing Hb A2 measurement in the Omani population. <i>Hemoglobin</i> , 2014 , 38, 299-302	0.6	4
11	A protein truncation test for Emery-Dreifuss muscular dystrophy (EMD): detection of N-terminal truncating mutations. <i>Neuromuscular Disorders</i> , 1999 , 9, 247-50	2.9	4
10	Broader spectrum of β thalassemia mutations in Oman: regional distribution and comparison with neighboring countries. <i>Hemoglobin</i> , 2015 , 39, 107-10	0.6	2

9	Multiple genomic aberrations in a patient with mental retardation and hypogonadism: 45,X/46,X,psu dic(Y) karyotype, thyroid hormone receptor beta (THRB) mutation and heterozygosity for Wilson disease. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 2231-5	2.5	2
8	Defining the proximal border of the Huntington disease candidate region by multipoint recombination analyses. <i>Genomics</i> , 1993 , 16, 599-604	4.3	2
7	Detection of truncated dystrophin in fetal DMD myotubes. <i>Advances in Experimental Medicine and Biology</i> , 1990 , 280, 17-23	3.6	2
6	DNA-Based Techniques for Detection of Carriers of Duchenne and Becker Muscular Dystrophy 2001 , 111-135		1
5	Clinical and Genetic Aspects of Hereditary Cerebral Hemorrhage with Amyloidosis Dutch Type (HCHW A-D) 2000 , 103-119		1
4	Duchenne and Becker muscular dystrophy. <i>Methods in Molecular Medicine</i> , 2004 , 92, 311-41		
3	Molecular Analysis of Facioscapulohumeral Muscular Dystrophy (FSHD1) 2001 , 305-316		
2	Somatic Triple Mosaicism in a Carrier of X-Linked Chronic Granulomatous Disease. <i>Blood</i> , 1998 , 91, 252-257		
1	Emerging Technologies, Need for Quality Assessment 2010 , 333-340		